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Terminologies and ontologies in biomedicine: Can text mining help?

Biomedical resources for text mining

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Overview

- An example
- Three types of resources
  - Lexical resources
  - Terminological resources
  - Ontological resources
- Some issues

An example

Neurofibromatosis 2

Neurofibromatosis 2

Neurofibromatosis type 2 (NF2) is often not recognised as a distinct entity from peripheral neurofibromatosis. NF2 is a predominantly intracranial condition whose hallmark is bilateral vestibular schwannomas. NF2 results from a mutation in the gene named merlin, located on chromosome 22.

[Uppal, S., and A. P. Coatesworth. “Neurofibromatosis Type 2.” *Int J Clin Pract*, 57, no. 8, 2003, pp. 698-703.]

Entity recognition

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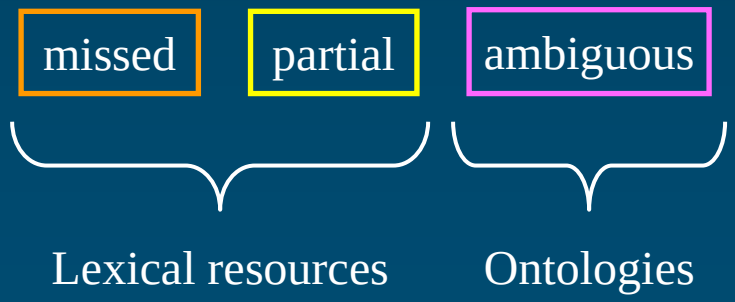


Relation extraction

Neurofibromatosis type 2 (NF2) is often not recognised as a distinct entity from peripheral neurofibromatosis. NF2 is a predominantly intracranial condition whose hallmark is bilateral vestibular schwannomas. NF2 results from a mutation in the gene named merlin, located on chromosome 22.

- vestibular schwannomas *manifestation of* neurofibromatosis 2
- neurofibromatosis 2 *associated with* mutation of NF2 gene
- NF2 gene *located on* chromosome 22

Ontologies

Resources for text mining

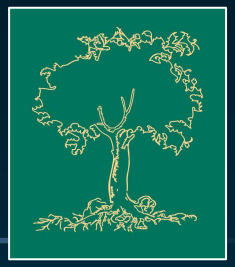
Types of resources

- Lexical resources
  - Collections of lexical items
  - Additional information
    - Part of speech
    - Spelling variants
  - Useful for entity recognition
  - UMLS SPECIALIST Lexicon, WordNet
- Ontological resources
  - Collections of
    - kinds of entities (substances, qualities, processes)
    - relations among them
  - Useful for relation extraction
  - UMLS Semantic Network, SNOMED CT

Types of resources (revisited)

- Lexical and terminological resources
  - Mostly collections of names for biomedical entities
  - Often have some kind or hierarchical organization (e.g., relations)
- Ontological resources
  - Mostly collections of relations among biomedical entities
  - Sometimes also collect names

Unified Medical Language System



- SPECIALIST Lexicon
  - 200,000 lexical items
  - Part of speech and variant information
- Metathesaurus
  - 5M names from over 100 terminologies
  - 1M concepts
  - 16M relations
- Semantic Network
  - 135 high-level categories
  - 7000 relations among them

Lexical resources

Terminological resources

Ontological resources

Lexical resources

SPECIALIST Lexicon

SPECIALIST Lexicon

- Content
  - English lexicon
  - Many words from the biomedical domain
- 200,000+ lexical items
- Word properties
  - morphology
  - orthography
  - syntax
- Used by the lexical tools

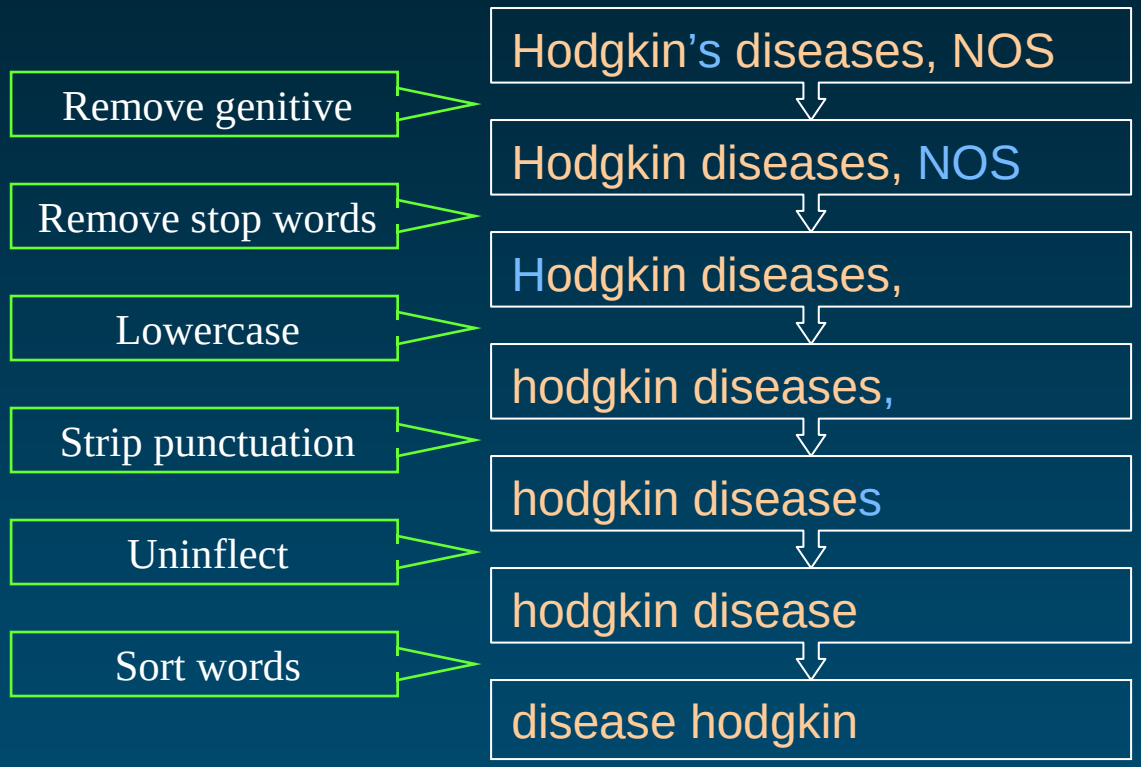
SPECIALIST Lexicon record

```
{
  base=hemoglobin      (base form)
  spelling_variant=haemoglobin
  entry=E0031208       (identifier)
  cat=noun             (part of speech)
  variants=uncount     (no plural)
  variants=reg         (plural: hemoglobins , haemoglobins)
}
```

Lexical tools

- To manage lexical variation in biomedical terminologies
- Major tools
  - Normalization
  - Indexes
  - Lexical Variant Generation program (lvg)
- Based on the SPECIALIST Lexicon
- Used by noun phrase extractors, search engines

Normalization



Normalization: Example

Hodgkin Disease
HODGKINS DISEASE
Hodgkin's Disease
Disease, Hodgkin's
Hodgkin's, disease
HODGKIN'S DISEASE
Hodgkin's disease
Hodgkins Disease
Hodgkin's disease NOS
Hodgkin's disease, NOS
Disease, Hodgkins
Diseases, Hodgkins
Hodgkins Diseases
Hodgkins disease
hodgkin's disease
Disease, Hodgkin

Normalization Applications

- Model for lexical resemblance
- Help find lexical variants for a term
  - Terms that normalize the same usually share the same LUI
- Help find candidates to synonymy among terms
- Help map input terms to UMLS concepts

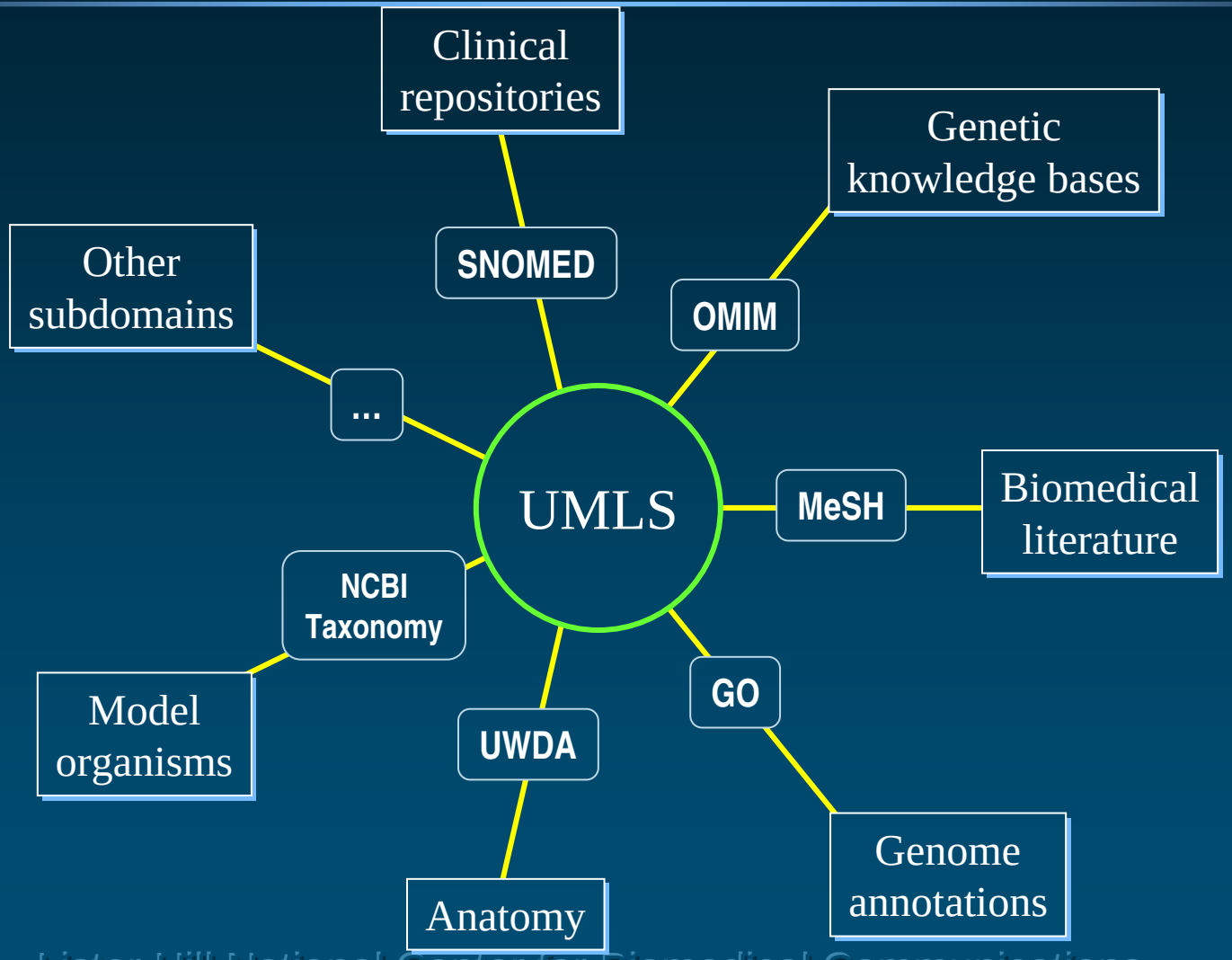
Terminological resources

UMLS Metathesaurus

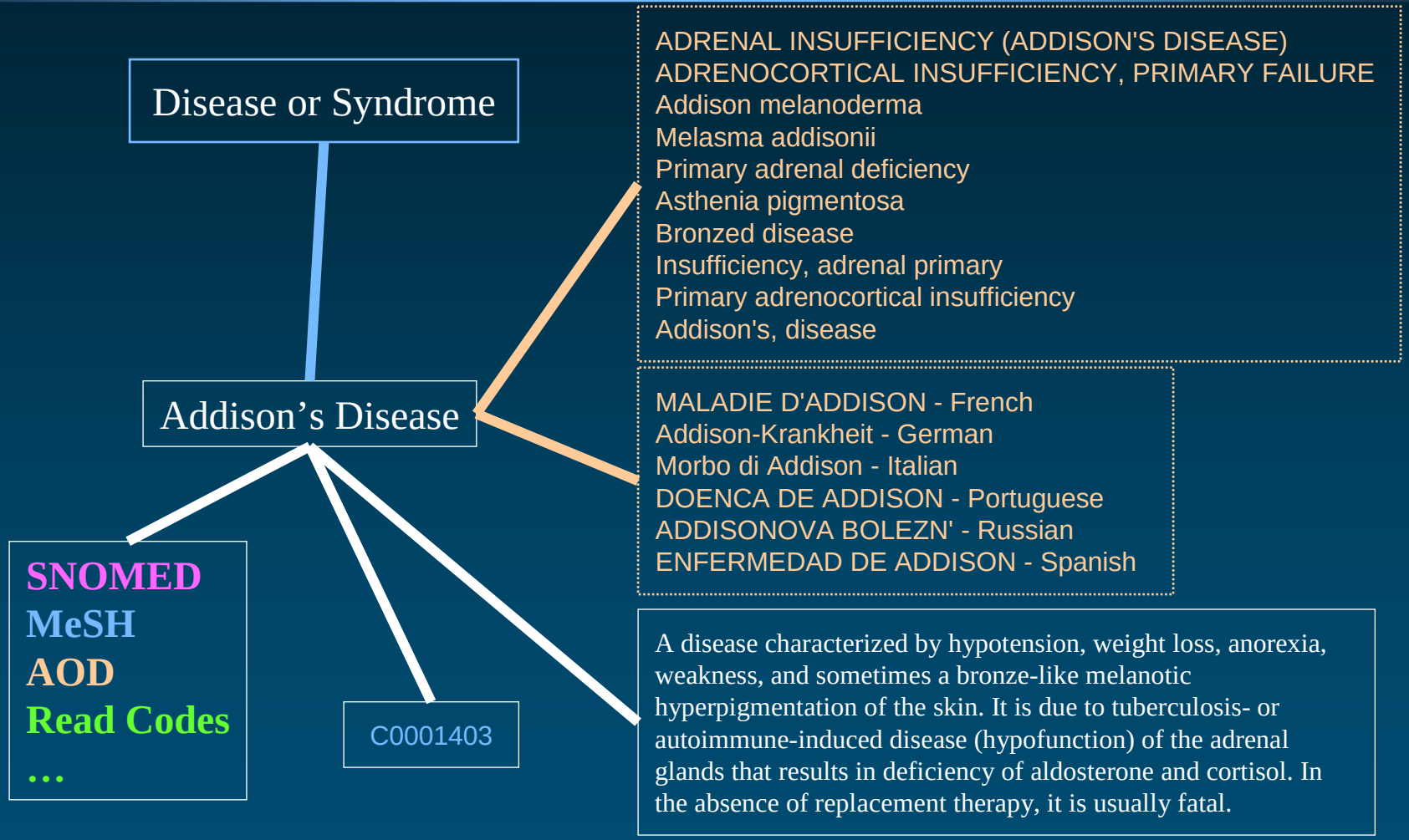
Source Vocabularies (2005AA)

- 134 source vocabularies
  - 132 contributing concept names
- Broad coverage of biomedicine
  - 5M names
  - 1M concepts
  - 16M relations
- Common presentation

Integrating subdomains

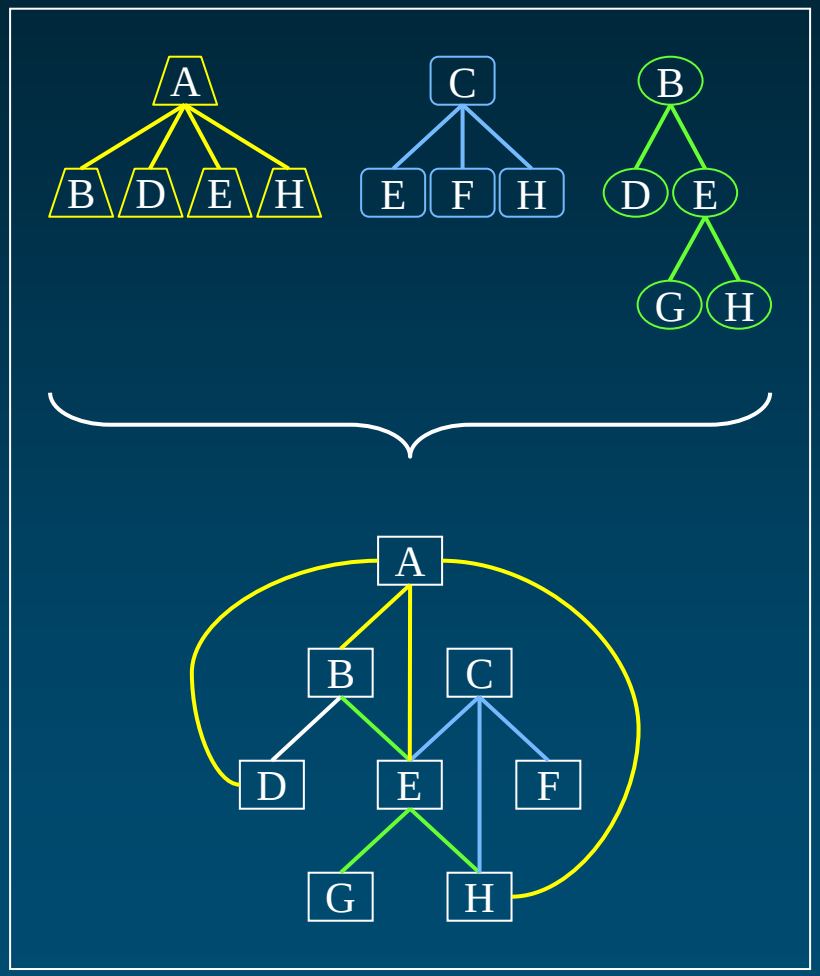


Addison's Disease: Concept



Organize concepts

- Inter-concept relationships: hierarchies from the source vocabularies
- Redundancy: multiple paths
- One graph instead of multiple trees (multiple inheritance)



Metathesaurus concepts Examples

Neurofibromatosis type 2	s	C0027832	Neurofibromatosis 2
NF2	s	C0085114	Neurofibromatosis 2 genes
peripheral neurofibromatosis	s	C0027831	Neurofibromatosis 1
[bilateral] vestibular schwannomas	a	C0027859	Neuroma, Acoustic
mutation / mutations	s	C0026882	Mutation
gene	s	C0017337	Genes
merlin	m	C0254123	Neurofibromin 2
chromosome 22	s	C0008665	Chromosomes, Human, Pair 22

Metahesaurus relations Examples

- Neurofibromin 2
  - Multiple parent concepts
    - Membrane proteins [MeSH]
    - Tumor suppressor proteins [MeSH]
    - Signaling protein [NCI Thesaurus]
  - 1 child concept
    - Merlin, Drosophila [MeSH]
  - Co-occurring concepts in MEDLINE
    - Neurofibromatosis 2 [13]
    - Membrane proteins [8]
    - …

Ontological resources

UMLS Semantic Network

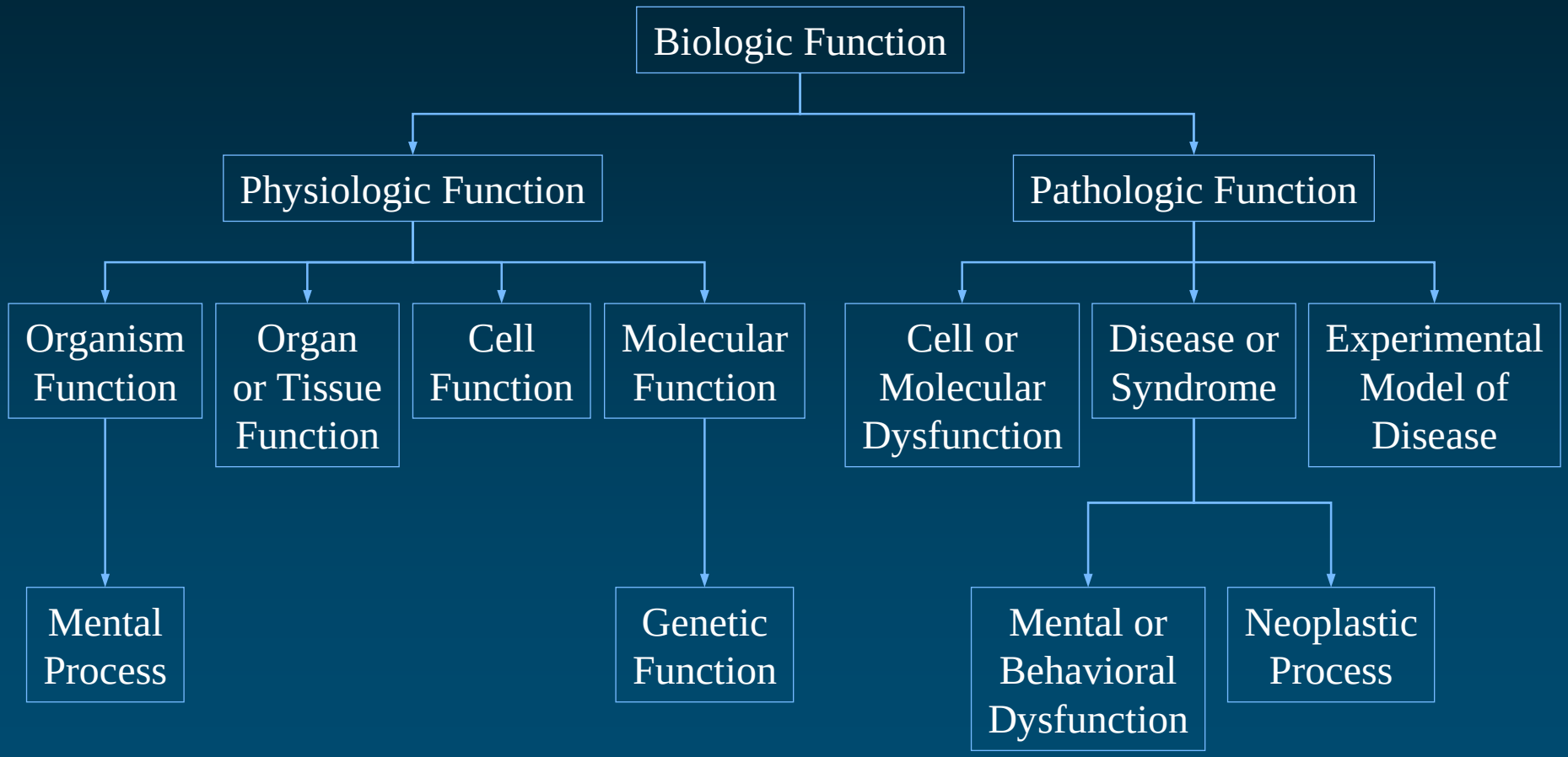
Semantic Network

- Semantic types (135)
  - tree structure
  - 2 major hierarchies
    - Entity
      - Physical Object
      - Conceptual Entity
    - Event
      - Activity
      - Phenomenon or Process

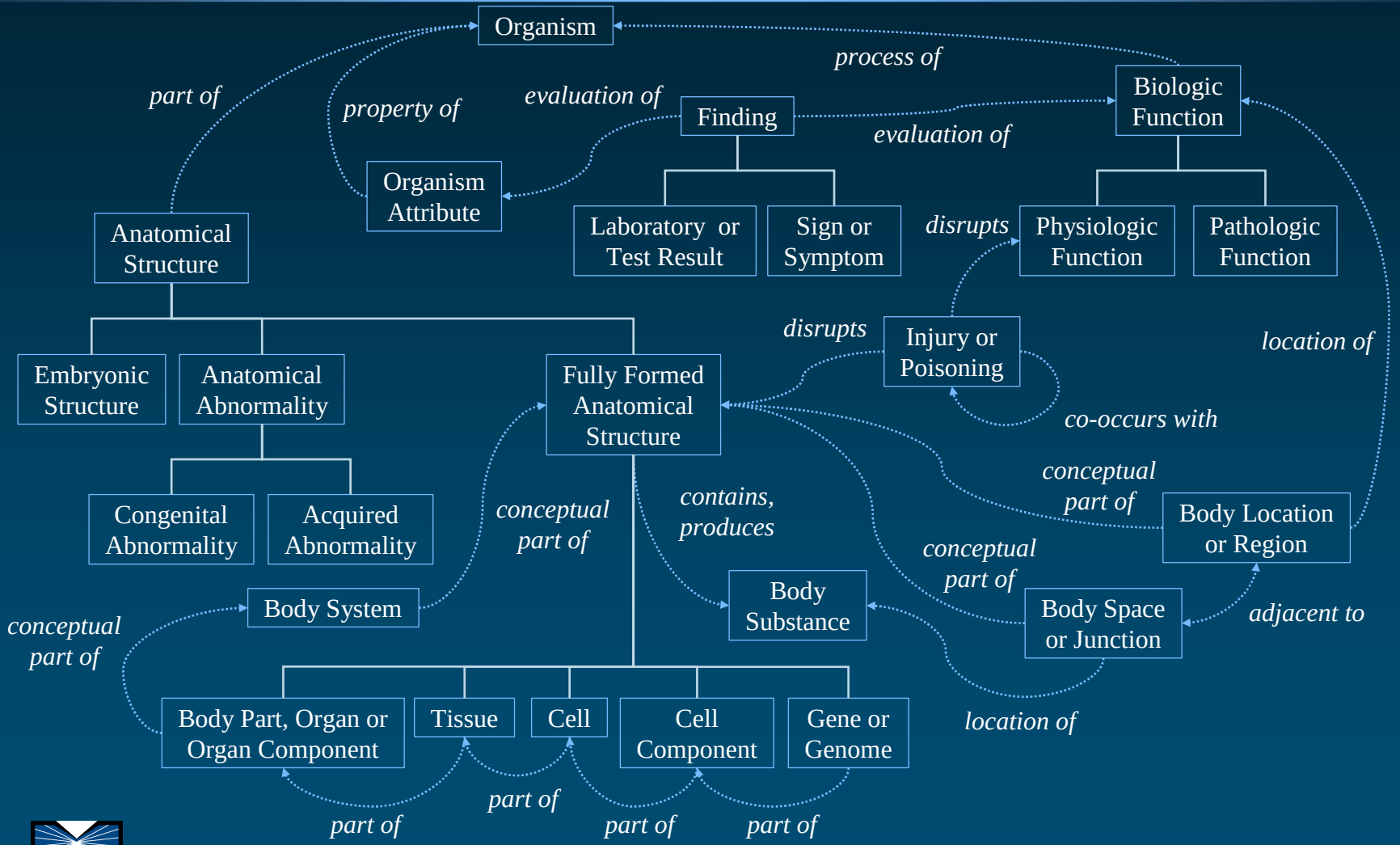
Semantic Network

- Semantic network relationships (54)
  - hierarchical (isa = is a kind of)
    - among types
      - Animal *isa* Organism
      - Enzyme *isa* Biologically Active Substance
    - among relations
      - treats *isa* affects
  - non-hierarchical
    - Sign or Symptom *diagnoses* Pathologic Function
    - Pharmacologic Substance *treats* Pathologic Function

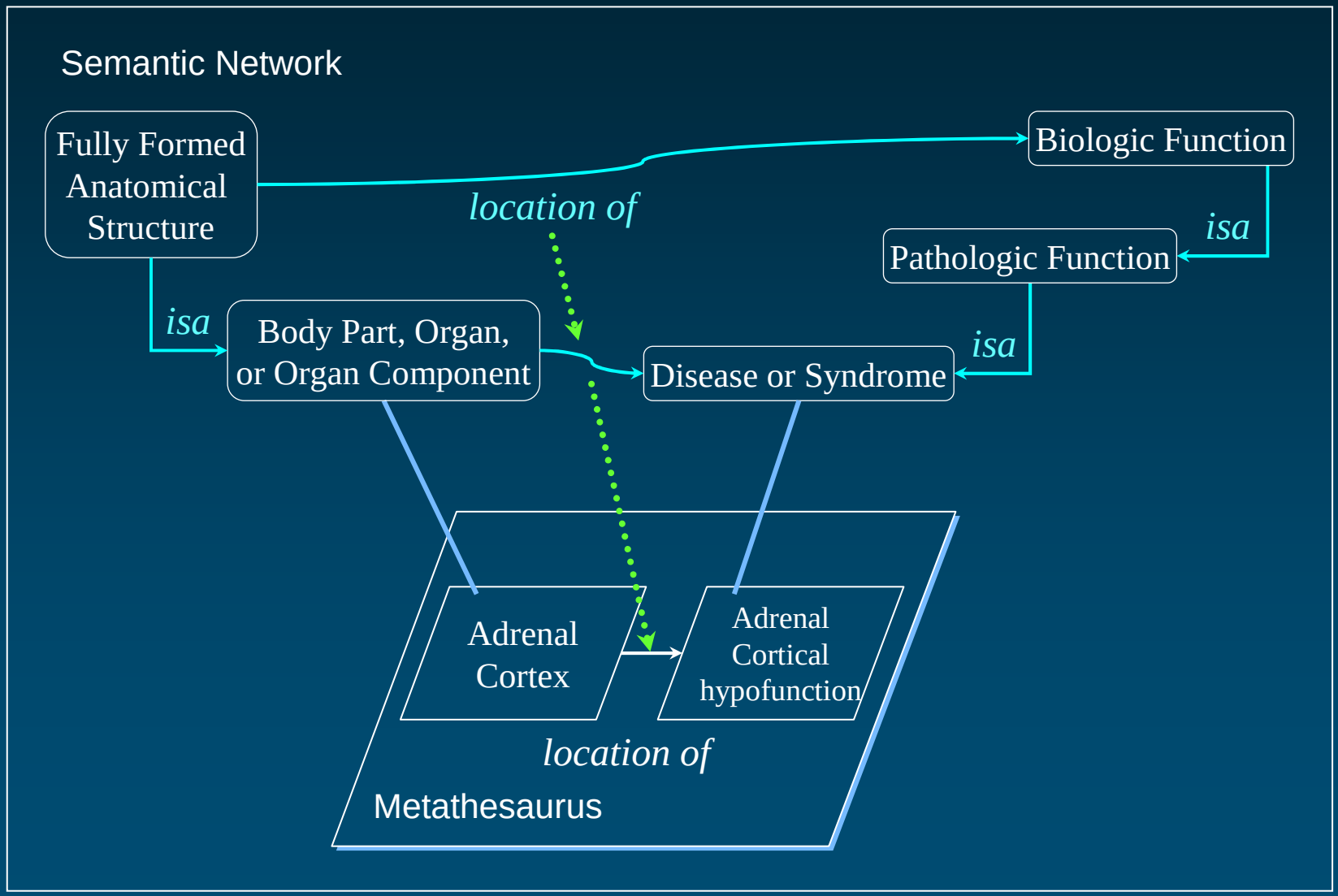
“Biologic Function” hierarchy (isa)



Associative (non-isa) relationships

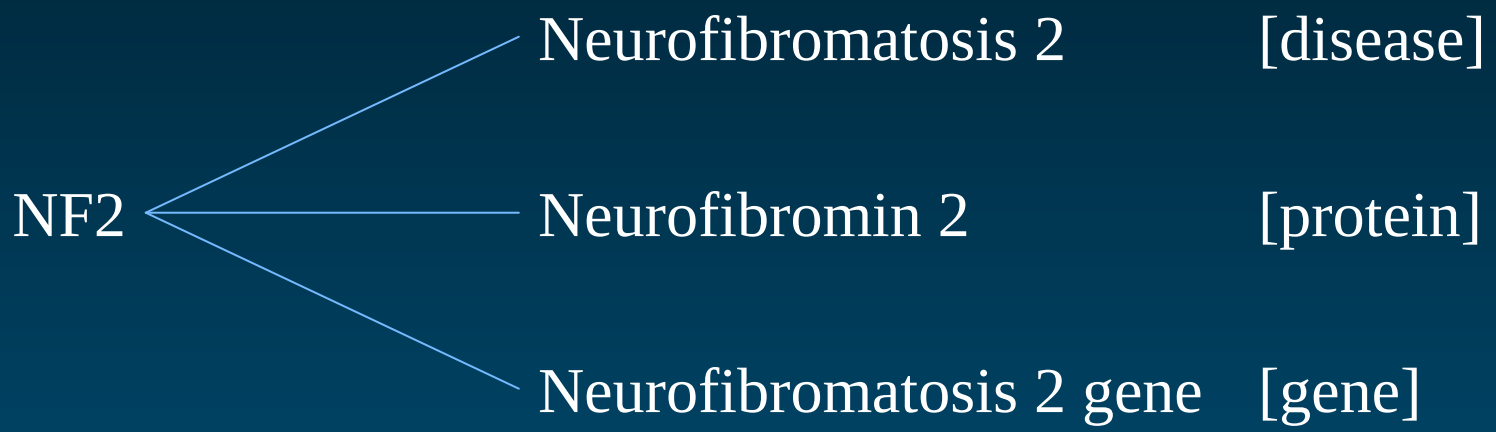


Relationships can inherit semantics



Some issues related to these resources

Ambiguity


NF2
Neurofibromatosis 2
[disease]
Neurofibromin 2
[protein]
Neurofibromatosis 2 gene
[gene]

Limited coverage

- e.g., Gene and protein names
  - Additional sources
  - Additional identification methods

Genew	http://www.gene.ucl.ac.uk/nomenclature/
Entrez Gene	http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=gene
UniProt	http://www.ebi.uniprot.org/index.shtml

Conclusions

Conclusions

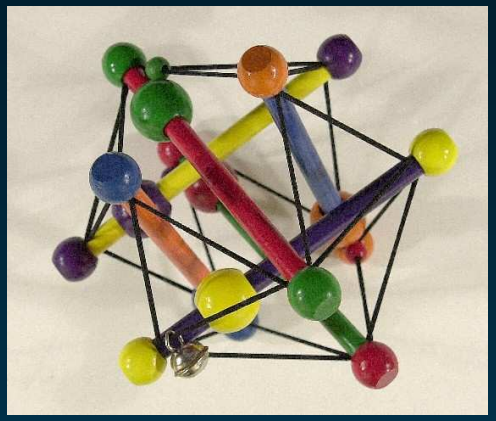
- Lexical and terminological resources enable entity recognition
- Terminological and ontological resources enable relation extraction

But…

- Text mining techniques can also benefit
  - Terminologies: term extraction
  - Ontologies: ontology population

UMLS documentation and support

- UMLS homepage http://umlsinfo.nlm.nih.gov/
  - with links to all other UMLS information
- UMLSKS homepage http://umlsks.nlm.nih.gov/
  - with links to the User's and Developer's guides
- Email address for support custserv@nlm.nih.gov



Medical Ontology Research

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